Impact of Children with Rare Diseases on the Family

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BACKGROUND

- Barth syndrome (BTHS) and Congenital muscular dystrophy (CMD) are rare diseases typically diagnosed during infancy or early childhood. Children with these rare diseases suffer from muscle fatigue and weakness [1; 2].
- Most previous studies have investigated the natural history and clinical features of children with BTHS and CMD, while only a few studies focused on parental caregiving issues.
- Evidence is sparse regarding the extent to which children with BTHS and CMD influence quality of life and family functioning of their parents. The provision of this evidence is important for understanding these parents' challenges and for planning effective healthcare and psychosocial services to meet their needs.

PURPOSE

- The purpose of this cross-sectional study is to **investigate how a child's functional ability, family cohesion, and satisfaction with healthcare affect quality of life and family functioning (family impact) for parents of children with rare diseases.**

METHODS

- Participants
  - Rare disease group
    - Parents of children with BTHS
    - Parents of children with CMD
  - Unaffected group
    - Parents of unaffected children
- Instruments
  - PedsQL™ Family Impact Module
  - Modified Barthel Index
  - Family Adaptability and Cohesion Evaluation Scale-IV
  - PedsQL™ Healthcare Satisfaction Generic Module
  - Demographic information form
- Data Analysis
  - Chi-square test
  - Independent samples *t*-test
  - Pearson correlation coefficient
  - Regression using the residual centering approach

Table 1. Demographic Information

	Rare Disease (N=40)	Unaffected (N=40)	*p*
Children			
Age			
Mean (*SD*)	10.97 (3.58)	10.83 (3.64)	.863
Actual Range	5 - 18	5 - 19	
Parents			
Age			
Mean (*SD*)	42.59 (6.16)	41.45 (6.44)	.424
Actual Range	29 – 53	29 - 54	

SD = standard deviation
*$p < .05$

This study was approved by the University of Florida's Institutional Review Board. Written informed consent was obtained from the parent.

RESULTS

Impact of Child, Family, and Healthcare Satisfaction on Parents of Children with Rare Diseases

	Family Impact		
	B	*SE B*	β
Child's Functional Ability	.197	.071	.410*
R^2	.168		
F	7.695*		

	Family Impact		
	B	*SE B*	β
Family Cohesion	1.496	2.745	.088
R^2	.008		
F	.297		

	Family Impact		
	B	*SE B*	β
Healthcare Satisfaction	.343	.128	.399*
R^2	.159		
F	7.204*		

SE = standard error.
*$p < .05$

Comparison between Two Groups

	Family Impact		
	B	*SE B*	β
Child's Functional Ability (MBI)	.207	.066	.356*
Group	-12.036	4.356	-.314*
MBI*Group	-1.519	.813	-.167
R^2	.391		
F	16.250*		

	Family Impact		
	B	*SE B*	β
Family Cohesion	3.753	1.910	.191
Group	-22.480	3.730	-.586*
Family Cohesion*Group	-5.771	3.914	-.138
R^2	.337		
F	7.204*		

SE = standard error.
*$p < .05$

- Both the child's functional ability and healthcare satisfaction were significantly related to quality of life and family functioning (family impact) for parents of children with rare diseases ($p<.05$); family cohesion was not related ($p>.05$).
- The impact of the child's functional ability and family cohesion on parental quality of life and family functioning (family impact) was not significantly different between the rare disease group and the unaffected group ($p>.05$).

CONCLUSIONS

- By investigating the factors that affect parents of children with rare diseases, this study expands knowledge regarding how a child's functional ability, family cohesion, and satisfaction with healthcare relate to parental quality of life and family functioning.
- Functional ability and satisfaction with healthcare are important factors affecting parental quality of life and family functioning in parents of children with rare diseases.
- Health professionals should develop effective strategies to support positive family relationships, daily living activities, and well-being for the children with rare diseases and their family members.

Reference
[1] Angelini, C., & Tasca, E. (2012). Fatigue in muscular dystrophies. *Neuromuscular Disorders, 22,* S214-S220.
[2] Mazzocco, M. M. M., Henry, A. E., & Kelly, R. I. (2007). Barth syndrome is associated with a cognitive phenotype. *Journal of Developmental and Behavioral Pediatrics, 28*(1), 22-30.